Gene Scene

Genetic consultation

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This article is the second in a series on the role of genetics in primary care. See this article on our web site (www.ewjm.com) for links to the other articles in the series.

INTRODUCTION

Patients view their primary care providers as the main source of genetic testing information. In 1998, the American Medical Association reported that 71% of persons surveyed would contact their primary care physician first if they had a question about a possible heritable disorder in their family.[1] Last month we focused on the uses of genetic testing and touched on the integral role of counseling in genetic testing.[2] This month we explore the elements of a genetics consultation to help primary care providers realize their possible roles when caring for patients with inherited diseases.

Cynthia, a 25-year-old woman who has been legally blind since age 14 as a result of a genetic condition, Stargardt disease, sees her physician for routine health maintenance. She indicates that she is getting married in a few months and is planning to have children, but is worried that they might also become blind. Her ophthalmologist had referred her for genetic counseling, but she never made an appointment because she did not want to travel the long distance to the nearest major metropolitan area only to be told what she could and could not do.

GENETIC CONSULTATION DEFINED

Genetic consultation consists of *genetic evaluation,* the process of information-gathering on a patient or family with a known or suspected genetic disorder, and *genetic counseling,* the process of helping patients understand the nature and cause of the inherited disorder and providing information that allows them to make informed medical and personal decisions.[3,4]

An individual or a family may seek a genetics consultation for several reasons:

- Confirming, diagnosing, or ruling out a genetic condition
- Identifying medical management issues
- Understanding genetic risks
- Obtaining psychosocial support

Genetic consultations are usually performed by trained genetics professionals working as a team that typically consists of a medical geneticist and genetic counselor or genetic nurse (see box).

Clinical genetics professionals

Medical geneticist: usually an individual who has obtained a doctorate (MD or DO, occasionally a PhD) and has completed an accredited 2-year fellowship in medical genetics. The American Board of Medical Genetics (ABMG) certifies medical geneticists

Genetic counselor: an individual who has completed an accredited master's level training program in genetic counseling. The American Board of Genetic Counseling certifies genetic counselors. (Before 1993, the ABMG certified genetic counselors)

Genetic nurse: typically a registered or master's level nurse with special training in human genetics

ELEMENTS OF A GENETICS CONSULTATION

Assessment

The genetics professional explores with the patient and family what they believe to be the reason for referral, their understanding of genetics and the diagnoses under consideration, their perception of disease status or risk, their beliefs about cause of disease, and their perception of disease burden. For infants and children, the patient's birth history is documented. For all patients, the medical history and current medical status are reviewed. A directed family history in pedigree form using standard symbols (see figure) is obtained, with emphasis on essential information (see box).[4,5]

Essentials of a genetic family history

- Information (age, health status, cause of death) on first-degree relatives (children, siblings, parents), second-degree relatives (grandparents, grandchildren, aunts, uncles, nieces, nephews), and more distant relatives as appropriate
- Status of current pregnancies
- Ethnic background
- Presence or absence of consanguinity

A psychosocial assessment is performed on all patients. This assessment involves the following:

- Review of the family's social history, education, employment, and social functioning
- Assessment of the family's community, religious, and family support systems
- Identification of potential ethical issues, such as confidentiality, insurability, discrimination, and nonpaternity

Additional medical records, including diagnostic testing, on the patient and affected family members, are reviewed as needed.

The medical geneticist performs a physical examination on the patient and other family members when indicated. In contrast to consultants in other medical specialties, medical geneticists often rely on examination of one or more relatives to establish the diagnosis in the patient or to confirm the disorder for which the patient is at risk.

Evaluation

After consulting relevant references, the geneticist discusses the possible diagnoses, sharing information about the condition when the diagnosis is clear and exploring issues of further evaluation and genetic testing.[2]

Communication

When a diagnosis is established or strongly considered, the genetics professional shares information about the condition in terms appropriate for the family's level of understanding and emotional status. Relevant issues are the expected course of the disease, possible treatments or interventions, and the underlying genetic cause, if known, including pattern of inheritance. In anticipation of the family's need to communicate genetic-risk information, the geneticist describes the risks to family members (parents, siblings, offspring, and others in the extended family) compared with risks in the general population. Local resources are identified for geographically distant relatives in need of further evaluation or genetic counseling. Reproductive options are discussed in a manner appropriate to the patient's personal beliefs and his or her need or request for the information.

Support

The genetics professional helps the family cope by providing the following support:

- Recognizes and discusses the emotional responses of family members to information given (such as shock, disbelief, relief, fear, guilt, sadness, shame, acceptance)
- Assesses signs that might indicate the need for further psychosocial support
- Listens to the family's story to understand their perspective
- Explores strategies for communicating information to others, especially family members who may be at risk
- Provides written materials and referrals to support groups, other families with the same or similar condition, and local and national service agencies (see box)

Consumer-oriented Internet resources for genetic diseases

Genetic Alliance (www.geneticalliance.org)

Family Village (www.familyvillage.wisc.edu/index.html)

Clinical Genetics site, University of Kansas (www.kumc.edu/gec/geneinfo.html)

NOAH: New York Online Access to Health (www.noah-health.org)

Follow-up

The genetics professional maintains ongoing communication with the patient and family by providing written documentation of the clinic visit (either as a copy of the clinic note or a personal letter) and by contacting the patient to assess level of understanding and response to decisions made. Families are encouraged to contact the clinic in the future for updated information, when considering pregnancy, or whenever questions arise.

LOCATING A GENETICS PROFESSIONAL

In a survey of primary care providers in a 4-state region, Hayflick and colleagues found that 1 in 4 internists had no knowledge of available genetic services.[6] Evaluation of an extended family may involve referral of geographically dispersed relatives to local genetics clinics throughout the United States. Medical geneticists and genetic counselors are often affiliated with large academic institutions or medical centers. Directories are available to help locate genetics service providers. An example of such a directory is the GeneTests Clinic Directory at www.genetests.org.

THE ROLE OF THE PRIMARY CARE PHYSICIAN

The role of the primary care professional depends on the practitioner's comfort level with genetic assessment and genetic counseling principles, experience with the particular disease, the nature of the patient's questions, and the availability of expert genetic help.[7] Like other medical specialists, genetics professionals are available for initial consultation and will recommend follow-up visits as appropriate.

Patients with inherited disorders have identified fundamental elements that they value most from their primary care providers.[8] These elements include:

- Offering an environment in which consumers and providers work together
- Having families be central to care
- Giving consideration of their condition in all aspects of care
- Recognizing that all stages of life are affected
- Allowing peer support

With her physician's reassurance that a genetics consultation will help answer questions rather than create new burdens, Cynthia meets with the medical geneticist and genetic counselor in the regional clinic held monthly in the town 30 minutes from her community. Her diagnosis is confirmed through record review prior to the clinic visit. She and her fiancé are relieved to learn that Stargardt disease is inherited in an autosomal recessive manner and although her children will be carriers, their risk for visual impairment is low. Cynthia is also pleased to know that her sister in a neighboring state, who also has Stargardt disease and similar concerns, can be seen in a genetics clinic near her home.

CONCLUSION

The elements of a genetic consultation rely on many aspects of traditional medical care but differ in that information may need to be gathered from and conveyed to multiple family members, psychosocial support and consideration of ethical issues are central issues, and educational materials and support group referrals are basic services.

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